WE CLAIM:

1. An isolated and purified nucleic acid molecule comprising a nucleic acid sequence selected from the group consisting of:

(a) the nucleic acid sequence in Figure 6;

(b) the nucleic acid sequence in Figure 6, wherein the thymine nucleotide at position 34,201 is replaced by a cytosine nucleotide;

(c) the nucleic acid sequence in Figure 6, wherein the guanine nucleotide at position 33,714 is replaced by a cytosine nucleotide;

(d) the nucleic acid sequence in Figure 6, wherein the thymine nucleotide at position 34,201 is replaced by a cytosine nucleotide and the guanine nucleotide at position 33,714 is replaced by a cytosine nucleotide;

(e) the nucleic acid sequence in Figure 7;

(f) the nucleic acid sequence in Figure 7, wherein the guanine nucleotide at position 2397 is replaced by a cytosine nucleotide.

2. An isolated and purified nucleic acid molecule according to claim 1, wherein the nucleic acid molecule has the nucleic acid sequence shown in Figure 6.

3. An isolated and purified nucleic acid molecule according to claim 1, said nucleic acid molecule having the nucleic acid sequence shown in Figure 6, wherein the thymine nucleotide at position 34,201 is replaced by a cytosine nucleotide.

4. An isolated and purified nucleic acid molecule according to claim 1, said nucleic acid molecule having the nucleic acid sequence shown in Figure 6, wherein the guanine nucleotide at position 33,714 is replaced by a cytosine nucleotide.

5. An isolated and purified nucleic acid molecule according to claim 1, said nucleic acid molecule having the nucleic acid sequence shown in Figure 6,

wherein the thymine nucleotide at position 34,201 is replaced by a cytosine nucleotide and the guanine nucleotide at position 33,714 is replaced by a cytosine nucleotide,

6. An isolated and purified nucleic acid molecule according to claim 1, wherein the nucleic acid molecule has the nucleic acid sequence shown in Figure 7.

7. An isolated and purified nucleic acid molecule according to claim 1, said nucleic acid molecule having the nucleic acid sequence shown in Figure 7, wherein the guanine nucleotide at position 2397 is replaced by a cytosine nucleotide.

8. An isolated polypeptide comprising the amino acid sequence in Figure 8.

9. An isolated polypeptide comprising the amino acid sequence in Figure 8, wherein the arginine at position 696 is replaced by a proline.

10. A recombinant vector comprising a nucleic acid molecule according to claim 1.

11. The recombinant vector according to claim 10, wherein said nucleic acid molecule is operably linked to an expression control sequence suitable for expression of said nucleic acid sequence in a host cell.

12. A host cell comprising the recombinant vector according to claim 11, wherein said host cell is selected from a group comprising a strain of E.coli, Pseudomonas, Bacillus subtilis, Bacillus stearothermophilus, or other bacilli, other bacteria, yeast, other fungi, insect cells, plant cells, or murine, bovine, porcine, human or other mammalian cells.

13. A method of producing a wild-type IKAP polypeptide, comprising:

(a) culturing a host cell transformed with a vector of claim 10 containing a DNA molecule encoding for a wild-type IKAP polypeptide in a cell culture medium under conditions whereby the IKAP polypeptide is expressed, and

(b) isolating the thus-produced wild-type IKAP polypeptide.

14. A method of producing a mutant IKAP polypeptide, comprising:

(a) culturing a host cell transformed with a vector of claim 10 containing a DNA molecule encoding a mutant IKAP polypeptide in a cell culture medium under conditions whereby the mutant IKAP polypeptide is expressed, and

(b) isolating the thus-produced mutant IKAP polypeptide.

15. A method of screening a subject to determine if said subject has a mutation associated with FD, comprising:

(a) providing a biological sample containing the DNA of the subject to be screened;

(b) detecting FD mutations in said biological sample.

16. The method according to claim 15, wherein the FD mutation is a T-C mutation at position 34,201 in the DNA sequence of Figure 6.

17. The method according to claim 15, wherein the FD mutation is a G-C mutation at position 33,714 in the DNA sequence of Figure 6.

18. The method according to claim 15, wherein the FD mutation is a T-C mutation at position 34,201 and a G-C mutation at position 33,714.

19. The method according to claim 15, wherein the FD mutation is detected by an allele-specific oligonucleotide hybridization assay.

20. The method according to claim 15, wherein the DNA from said biological sample is amplified using oligonucleotide primers flanking the mutation.

21. The method according to claim 20, wherein the DNA is amplified with oligonucleotide primers 18F and 23R.

22. The method according to claim 20, wherein the DNA is amplified with oligonucleotide primers 19F and 23R.

23. The method according to claim 20, wherein the DNA is amplified with oligonucleotide primers 18F, 19F and 23R.

24. The method according to claim 23, wherein the amplified DNA is screened for FD mutations using an allele-specific oligonucleotide hybridization assay.

25. The method according to claim 24, wherein the hybridization assay is accomplished using probes that span the T-C mutation at nucleotide position 34,201 in the IKBKAP gene.

26. The method according to claim 24, wherein the hybridization assay is accomplished using probes that span the G-C mutation at nucleotide position 33,714 in the IKBKAP gene.

27. The method according to claim 24, wherein the hybridization assay is accomplished using probes selected from the following sequences:

(a) 5'- AAGTAAG(T/C)GCCATTG- 3', and

(b) 5'- GGTTCAC(G/C)GATTGTC- 3'.

28. The method according to claim 15, wherein the FD mutation is detected by method selected from the group consisting of:

(a) restriction-fragment-length-polymorphism detection based on allele-specific restriction-endonuclease cleavage,

(b) hybridization with allele-specific oligonucleotide probes including immobilized oligonucleotides or oligonucleotide arrays,

(c) allele-specific PCR, mismatch-repair detection (MRD),

(d) binding of MutS protein,

(e) denaturing-gradient gel electrophoresis (DGGE),

(f) single-strand-conformation-polymorphism detection,

(g) RNAase cleavage at mismatched base-pairs,

(h) chemical or enzymatic cleavage of heteroduplex DNA,

(i) methods based on allele specific primer extension,

(j) genetic bit analysis (GBA),

(k) oligonucleotide-ligation assay (OLA),

(l) allele-specific ligation chain reaction (LCR)

(m) gap-LCR, and

(n) radioactive and/or fluorescent DNA sequencing.

29. A kit for assaying for the presence of an FD mutation in an individual comprising at least one oligonucleotide probe capable of detecting the FD1 mutation or the FD2 mutation.

30. A kit according to claim 29, further comprising primers capable of amplifying the region containing said mutations.

31. A kit according to claim 30, wherein said primers are 18F and 23R.

32. A kit according to claim 30, wherein said primers are 19F and 23R

33. A kit according to claim 30, wherein said primers are 18F, 19F and 23R.

34. A kit according to claim 29, further comprising an oligonucleotide probe which specifically hybridizes to one or more additional mutant or wild-type genes, wherein said additional gene codes for a protein associated with an additional genetic disease.

35. A kit according to claim 34, wherein the additional genetic disease is selected from the group comprising: Canavan's disease, Tay-Sachs disease, Goucher disease, Cystic Fibrosis, Fanconi anemia, and Bloom syndrome.

36. A method of detecting a FD mutation in a sample, comprising isolation of RNA from a tissue sample, amplifying the RNA using primers in exons 19 and 23; determining whether said sample contains a mutant product or a wild-type product, wherein the identification of a mutant product indicates the presence of an FD mutation in said sample.

37. The method according to claim 36, wherein said RNA is isolated from neuronal tissue.

38. A method of detecting a FD mutation in a sample, comprising the utilization of an antibody capable of detecting a truncated protein product that is indicative of FD.

39. A method of producing a transgenic animal expressing a mutant IKAP mRNA comprising:

(a) introducing into an embryonal cell of an animal a promoter operably linked to the nucleotide sequence containing a mutation associated with FD;

(b) transplanting the transgenic embryonal target cell formed thereby into a recipient female parent; and

(c) identifying at least one offspring containing said nucleotide sequence in said offspring's genome.

40. The method according to claim 39, wherein said mutation is the FD1 mutation.

41. The method according to claim 39, wherein said mutation is the FD2 mutation.

42. The method according to claim 15, further comprising a determination of whether said individual is homozygous or heterozygous for said mutation.

43. An oligonucleotide for detecting a mutation associated with FD, said oligonucleotide having a sequence selected from sequences which detect an FD mutation or bind to a region flanking said FD mutation.